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A STUDY IN HEREDITY.

Thomson writing on heredity says: - "There are no scientific problems of greater human interest than those of heredity." - it might very truly be added, that few are of equal importance. More especially is this the case when we view the situation from the standpoint of the Physician; for do we not now recognise a close asso--ciation between the study of heredity and that of many diseases in so far as a predisposition to certain patho--logical conditions is certainly inherited.

It must indeed be acknowledged that in the depart--ment of science there remains much to be observed, and that the views of those who have specially studied this subject by no means correspond: none the less the study of heredity is, at length, surely if slowly emerging from that state of chaos in which, until comparatively recently, fact and fiction were inextricably blended; and as time progresses, we will, without doubt, be enabled to apply our knowledge of the subject, more and more usefully in the practice of medicine.

On this account it seems to me to be advisable, that true and complete records should when available and possible, be made of families, and it has been my good fortune to have come across a family whose physical peculiarities render it comparatively easy to trace.

Leaving for the present the nature of the defor--mities found, I wish first to draw attention to the

genealogy of the family, which appears to me to be much the more interesting of the two subjects.

There is of course in this as in most other cases of the kind the time-honoured, if unsupported, explanation of "maternal impression" as the cause of the deformity.

The history is shortly the following.

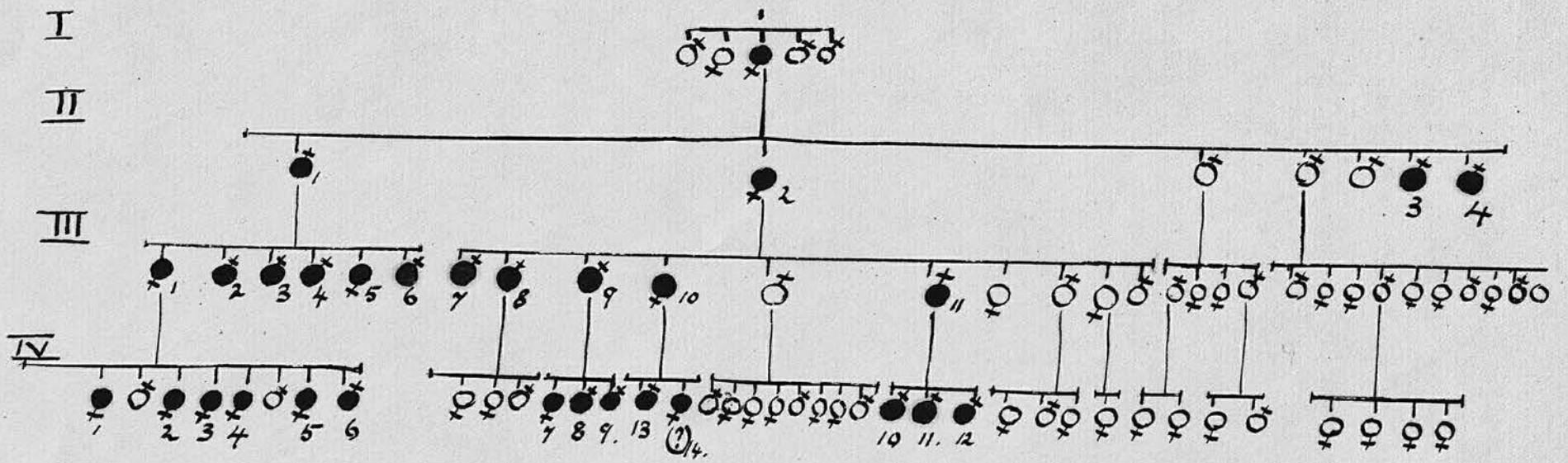
- The great-great-grandmother of the latest generation being pregnant happened to have committed a theft, when charged with the offence she denied it and further invoked her Creator to let her child when born have no fingers to steal with, should she be guilty. In due course the infant was born without fingers or toes and the woman confessed to the theft lest a greater evil might befall her.

- All very picturesque but equally unconvincing. As far as one can gather the woman was well advanced in pregnacy, neither is it explained by what process the digits were separated nor what was their ultimate fate.

The condition should rather be looked upon as due to an inherent lack of developmental vigour transmitted from one generation to another.

The first individual to show the deformity was the female child in question and in the second generation we find that she had a family of seven, three males and one female affected, and three males normal. Of the affected members of the family two were married, a male whose hands and feet were deformed, and a female whose feet alone showed the peculiarity.

Genealogical Tree.



● - deformed.
○ - Normal

No. IV. 14. whether deformed or otherwise uncertain probably deformed

Nos. II. 1, 3
III, 1, 3. 7. 11
IV 1, 2, 3, 4, 5, 6, 12, 13, 14
} all show same type of deformities, Hands narrow with 4th finger only present. feet only having 5th toe. or rarely, 1st or 5th

II. 2
III 2, 4, 5, 6, 8
IV. 7, 8. 9.
} show irregular deformities.

II. 4
III. 9. 10.
IV. 10. 11
} nature of deformities not known.

In the family of the male (James Sadler) there were six children, four males and two females, all having both hands and feet deformed: while Eliza Sadler (Mrs. Whitehorn) gave birth to ten children, four males and one female being deformed in both hands and feet while three males and two females were normal.

In the fourth generation we find five families showing deformities.

The family of

			deformed.	normal.	
(I)	1.III	giving	5 males & 1 female	2 males	= 8
(2)	" "	" 8.III "	0	2 males & 1 female	= 3
(3)	" "	" 9.III "	2 males & 1 female	0	= 3
(4)	" "	"10.III "	1 male and 1 female(?) (Foot note)	0	= 2
(5)	" "	"11.III "	3 males	0	= 3

Now there are two laws applying to heredity which have been proved to hold true in many cases.

Galton's law of ancestral inheritance, and, The law of dominants and recessives formulated by Gregor Johann Mendel, in 1866.

The former of these is of little importance in so far as cases of this sort are concerned. In it he states - That in a given generation the average heritage is

Footnote:- (There is a doubt as to whether the second child of 10.III is deformed or otherwise, probably the former.

made up of ancestral contributions, ½ being parental ¼ grand parental, 1/8 great-grand parental - and so on "ad infinitum."

Galton intended this law to apply to large numbers, and not to individuals, and while this fact greatly detracts from the value of its application in this specific instance the numbers being too small, nevertheless it is of interest to note that the result obtained approximates to that which one might expect.

For instance - If the third generation be taken we find in the case of two of the families one parent and one grand parent affected the number of individuals in the said fam--ilies being sixteen (16) while in the two remaining fam--ilies numbering thirteen (13) one grand parent alone is affected.

That is to say the members deformed in this generation should number $\left\{\left(\frac{1}{4}+\frac{1}{8}\right)\times 16\right\}+\left(\frac{1}{8}\times 13\right)$

$$= \frac{48+13}{8} = \frac{61}{8} = 8 \text{ (about).}$$

Actually we find eleven members affected, which is too large a number; but when we deal with a larger number of individuals as in the fourth generation, we find a much nearer approach to Galton's formula.

Here we find:-

(1) 5 families numbering nineteen having one parent, one grandparent and one great-grandparent affected.

(2) 3 families numbering twelve having one grand-parent and one great-grandparent affected.

and

(3) 3 families numbering eight having one great-grand-parent only affected.

Therefore it might be expected that the total of the deformed members would be : -

$$\left\{\left(\frac{1}{4}+\frac{1}{8}+\frac{1}{16}\right)\times 19\right\}+\left\{\left(\frac{1}{8}+\frac{1}{16}\right)\times 12\right\}+\left(\frac{1}{16}\times 8\right)$$

$$=\left\{\frac{7}{16}\times 19\right\}+\left\{\frac{3}{16}\times 12\right\}+\left\{\frac{1}{16}\times 8\right\}$$

$$=\frac{133+36+8}{16}=\frac{177}{16}=11 \text{ (about)}$$

Actually we find either 13 or 14 of the individuals showing deformities, depending on whether the second child of III (10) was deformed or otherwise.

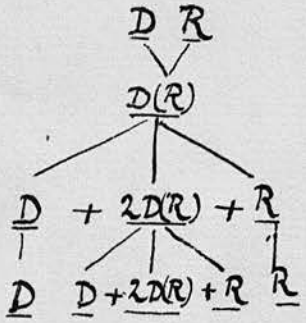
It is probable too that some of the other normal members of the third (3rd.) generation are married and have families, as I have been unable to discover where the majority have disappeared to, some I know are married and have moved out of this district, but it is instructive to note that in this generation six (6) out of the eleven deformed members are married, five having families, while I only know of seven of the eighteen normal members married, and six having families.

It is very improbable that natural selection would permit the least fitted members of the community to multiply to a proportionately greater extent those better able to take part in the struggle for existence, hence it might be assumed that there are a large number of normal children in the fourth generation, who are children of normal parents in the third generation in which case the number of <u>affected</u> members of the fourth generation according to <u>Galton's Law</u> instead of working out at $\frac{177}{16}$ would probably approach a much larger number.

In any case the result obtained is approximately correct though it is very possibly only a coincidence.

To turn to the law of recessives and dominants, which deals with individuals families and not generations, Mendel there formulates the theory that when two individuals of markedly different characters are paired

all the progeny resemble the dominant parent, but, when this progeny are further interbred we obtain $\frac{3}{4}$ of the total offsprings resembling the dominant type while $\frac{1}{4}$ show the recessive characters of the grand-parent. Furthermore of the $\frac{3}{4}$ showing dominant characters $\frac{1}{3}$ are pure dominant and when <u>interbred</u> breed true to their dominant characters while $\frac{2}{3}$ are impure dominants their progeny being again in the proportion of 3 showing dominant characters and 1 showing recessive characters. - The recessives breeding true as do the <u>pure</u> dominants.



The above graphic formula explains the law

D being pure dominants

D(R) " Impure "

R " pure recessive.

In this specific instance it must be admitted that in all probability the deformity is a dominant character, appearing as it does with the utmost regularity in each generation. In any case if the deformity be taken as the dominant character and the normal as the recessive we find a much closer approach to mendelian proportions than are usually found in the human species.

The weak point is in the first generation. If the first individual showing the deformity was a pure domi--nant then it might be expected that all her progeny would show the dominant character of deformity; but we find only four out of seven showing it.

The question naturally arises - Was the first case a pure dominant ? In the first place there is no previous history of deformity in the family, though of course it is going a long way back, and the history is very vague. Hence we might consider it a case of "spontaneous variation," but a variation embodying a dominant character. Now is it not possible that a variation might arise of an impure dominant type ?

It is instructive to note however that there is at least one other family in Berkshire showing a similar deformity also dominant in that it occurs in several generations. I have this information on good authority. The family living in the neighbourhood of Swindon, not a great distance from Wallingford, where the family in question originally came from. So of course the possibility arises that the deformity was directly transmitted from parent to offspring.

In any case if we look on the first case as an impure dominant paired to a pure recessive we should expect to find the progeny $\frac{1}{2}$ impure dominants and $\frac{1}{2}$ pure recessives.

Thomson demonstrated this by the following formula -

If $n\,\mathcal{D} + n\,\theta$ = egg-cell of impure dominant, be fertilised
by, $n\,\theta + n\,\theta$ = Sperm- " " pure recessive.

The result will be

$n\,\mathcal{D}$ fertilised by $n\,\theta = n\,\mathcal{D}$
and $n\,\theta$ " " $n\,\theta = n\,\theta$

That is to say equal numbers of impure dominants and pure recessives.

Now taking the affected families in successive generations we find

No. of families.	affec-ted.	Nor-mal.	(2) A	(2) N	(3) A	(3) N	(4) A	(4) N	(5) A	(5) N	Totals. affected.	Totals. Normal.
II Generation.	4	3									4	3
III "	6	0	5	5							11	5
IV "	6	2	0	3	3	0	2	0	3	0	13or14	5 or 6
											29	13
											or 28	14

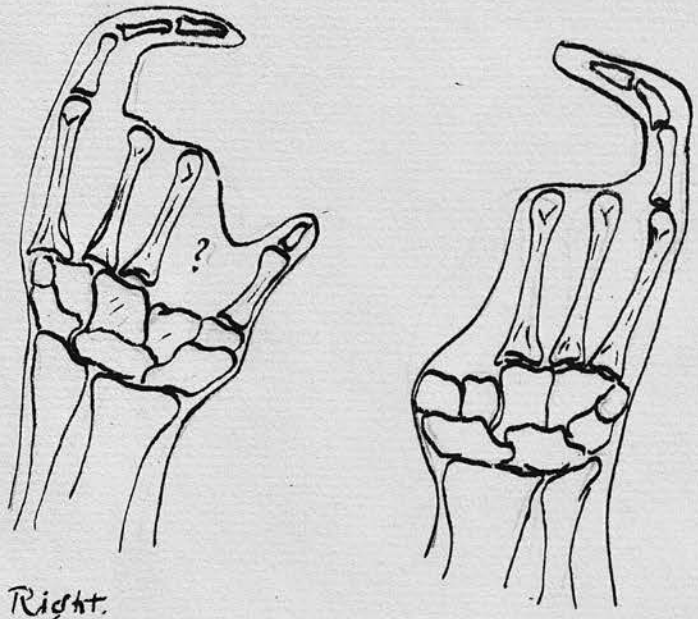
depending on whether the second child of 10.III was deformed or otherwise.

It is thus evident that approximately $\frac{2}{3}$ of the offsprings of the affected families are themselves deformed. Such a result approaches nearly enough the proportions necessary to demonstrate Mendelian phenomena when one considers the uncertainties of human existence, and a further confirmation of the proposition is found when one considers the fact that every recessive in the family has 'bred true.'

The deformities met with in the family are themselves not devoid of interest. As will be noted they vary considerably in different individuals, though a certain similarity is present between them.

In the majority of cases the outer portion of the hand is deficiant and the middle portion of the foot.

I have made out as well as I could, the bones present and those absent, by palpation. Such a method of course leaves much to be desires, but the country practitioner has not access to X Ray apparatus, and further many of the affected persons feel their affliction keenly and are not willing to have it further investigated.



John. Sadler.

II. 3.

II. (1) James Sadler: farm labourer: (dead) said to have had both hands and feet deformed. The 4th. finger only being present in the case of the hands while feet had the first and fifth toes present on each: (married, six children.)

(2) Eliza Sadler - Mrs. Whitehorn. (dead) - In this case the hands were said to be normal, the feet having only one toe, the smallest, present. (Married, ten children.)

(3) John Sadler, - scavenger - hands and feet deformed. (unmarried.)

Hands.	Right.	Left.
Carpals.	Apparently normal.	Apparently normal.
Metacarpals.	1st, 3rd, 4th, & 5th present. 2nd absent or rudimentary.	3rd, 4th & 5th present.
	4th finger 3 phal-anges.	4th Finger 3 phalanges present.
	Thumb. Rudimentary, one phalanx.	

Feet.

Examination refused, but said he had the 5th. toe only present on each foot.

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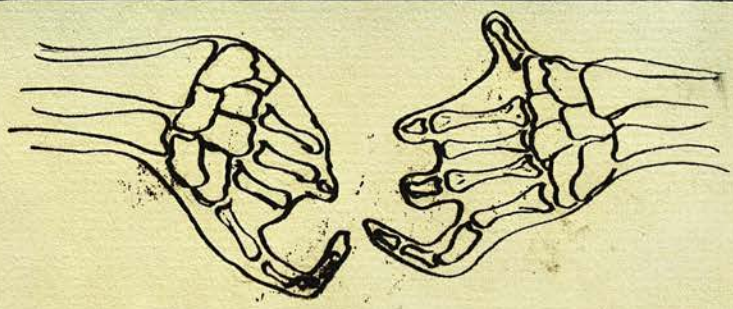
(4). George Sadler, (dead) hands and feet said to be deformed, no further details obtainable.

Richard. Sadler. III 2



Left foot.
Plantar. Surface

III. (1) Mrs. Richins, a female pauper, married, 8 children, hands said to be narrow and to have only the 5th digit present. Feet said to have 1st. and 5th toes present.

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(2) Richard Sadler. Inmate of workhouse, unmarried: hands and feet deformed.

Hands.

	Right.	Left.
Carpals.	Trapezium probably absent.	Normal.
Meta-carpals.	2nd, 3rd, 4th & 5th, present.	1st, 2nd, 3rd, 4th & 5th present. 1st rudimentary.
Phalanges.	Mid finger 1 rudimentary phalanx. 4th. finger 3 phalanges.	first finger, one rudimentary phalanx. Ring finger, one fairly well-developed phalanx. Little finger, 3 phalanges.

Feet, both show the same deformity.

Tarsals, probably mid, and external cuneiform bones absent.

Meta-tarsals. 1st and 5th only present.

Phalanges. 1st toe. 2 fused phalanges.
5th toe. 3 fused phalanges.
(i.e. no moveable joints are demonstrable.)
A web of skin is present between the two digits.

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(3) Francis Sadler - farm labourer. Hands said to have one finger only, the 4th present.

Feet said to resemble last case.

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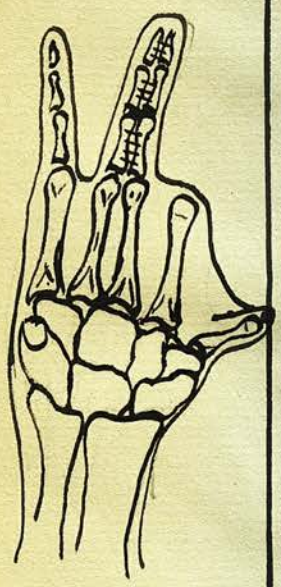
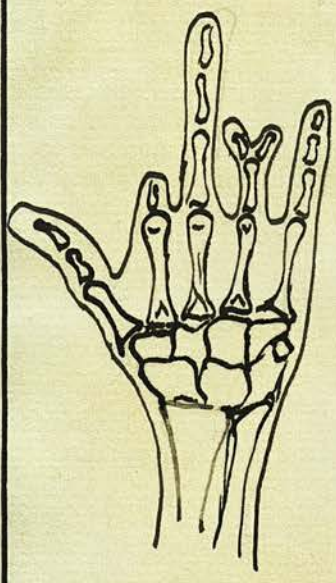
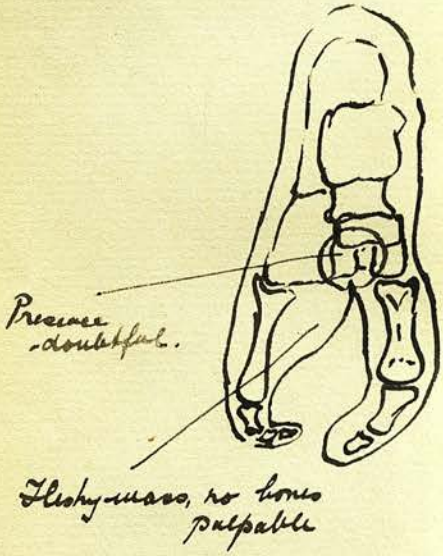
(4) - Sadler, dead, unmarried. Hands and feet said to be deformed: deformities irregular.

Eliza. Sadler (Mrs Franklin)

III. 5







III. (5) Eliza Sadler. (Mrs. Franklin.) married - no living children: has had two abortions at the 4th. month. Has both hands and feet deformed.

Hands.

	Right.	Left.
Carpals.	Normal.	Normal.
Meta-carpals.	Normal.	Normal.
Digits.	Thumb, one very rudimentary bone. Index finger absent. Mid and ring fingers united, one nail only present, and bones partially fused. 3 phalanges to each finger. 4th finger, normal.	Thumb, short otherwise normal. 1st finger, one small rudimentary phalanx. Mid finger, very long 3 phalanges. Ring finger dichotomous at 2nd joint. 4th finger, normal.

Feet.

Both show a similar deformity.

Presence of mid. and Ext. cuneiform bones doubtful.

1st and 5th Meta-tarsals only present.

Phalanges, normal in number, but fused.

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(6) Henry Sadler, farm labourer, unmarried.

Hands and feet deformed.

Hands.

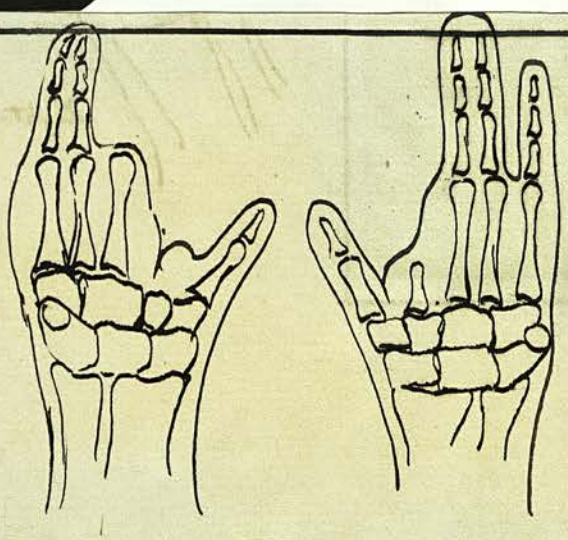
	Right.	Left.
Carpals.	Apparently normal.	Apparently normal.
Meta-carpals.	1st, 3rd, 4th and 5th present.	1st, 2nd (probably small & rudimentary 3rd, 4th and 5th.
Digits.	Thumb, rudimentary, one small phalanx. Ring & 5th fingers united by thick fleshy web. 3 phalanges to each.	Thumb, rudimentary, one small bone. Middle and ring fingers fused, one nail. 3 phalanges to each. 5th finger, normal.

Feet.

Show a deformity similar to Eliza S - . III (5)

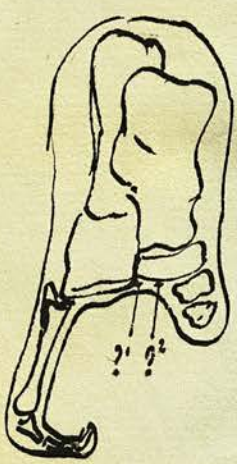
Henry Sadler.

III . 6.



Thomas Whitehorn.

III . 11



III. (7) Wm. Whitehorn, dead, unmarried: hands and feet said to have been deformed.

Said to have had only the little finger and 5th toe present.

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(8) Alfred Whitehorn, married. (3 normal children) hands and feet said to be deformed. Hands said to be broad with a thumb on right hand, and feet to have 5th toe only present.

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(9) Chas. Whitehorn, (married) whereabouts unknown, (has 3 deformed children). Hands and feet in this case said to be affected; in what manner not specified.

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(10) Lizzie Whitehorn, (flower seller) said to have hands and feet affected, has 2 children, one certainly showing deformity, the other doubtful.

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(11) Thomas Whitehorn, (Green keeper on Golf course) married, 3 children showing deformity.

Hands.

Right & Left.

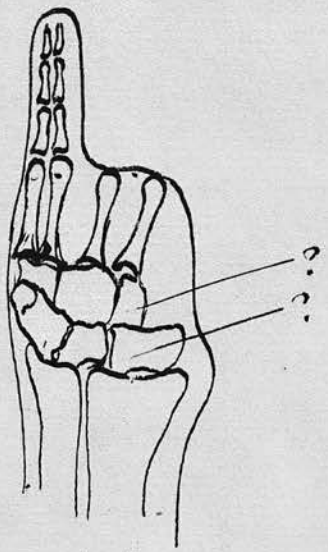
Carpals.	Trapezium presence doubtful. Trapezoid probably not fully-developed on left hand.
Metacarpals.	3 inner bones alone present.
Digits.	4th finger only present, very much curved, 3 phalanges.

Feet.

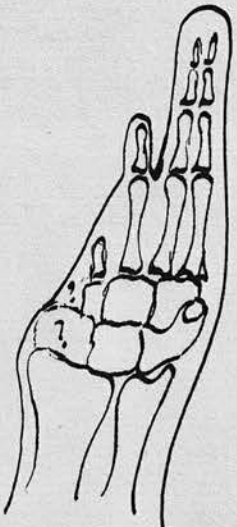
Tarsus.	Presence of mid and external cuneiforms doubtful.
Metatarsals.	5th alone present. A small hard mass is present lying over internal cuneiform bone, possibly a rudimentary 1st metatarsal.
Digits.	5th alone present, much curved. 3 phalanges.

Eliza. Whitehorn.

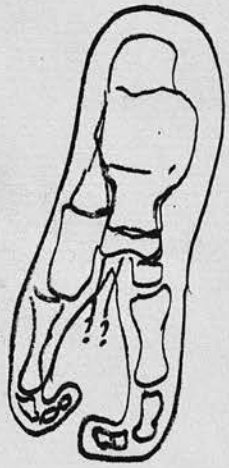
IV. 7.



Right. Hand.



Left. Hand.



Right foot.

IV. (1) (2) (3) (4) (5) female children of III.(1) Mrs. R - all said to show deformities of hands and feet, the hands said to be very narrow and to have one finger the 4th, alone present on every case., while the feet had the 5th toe alone present.

Three of these children were in the Cottage Homes at Wallingford several years ago and my information regarding them was obtained from the Medical Officer at that time, Mr. E. Horne.

(6) Male child (- R) Still Born, hands and feet as above.

(7) Eliza Whitehorn. Hands and feet deformed.

Hands.

	Right.	Left.
Carpus.	Scaphoid presence doubtful. Trapezium probably absent.	Scaphoid and Trapezium probably absent.
Metacarpals.	2nd, 3rd, 4th, and 5th present.	2nd (Rudimentary) 3rd, 4th and 5th.
Digits.	Ring and little fingers closely united by fleshy web, double nail. 3 bones in each.	Ring and little fingers closely united by a thick fleshy web, double nail. 3 phalanges in each finger. Rudimentary middle finger, one small phalanx.

Feet.

Same deformity in each foot.

Tarsus, presence of the middle and ext. cuneiform bones doubtful.

Metatarsals. 1st and 5th only present.

Digits. 1st and 5th alone present, much curved, resembling a crab's claws, normal number of bones in each digit, but joints are ankylosed.

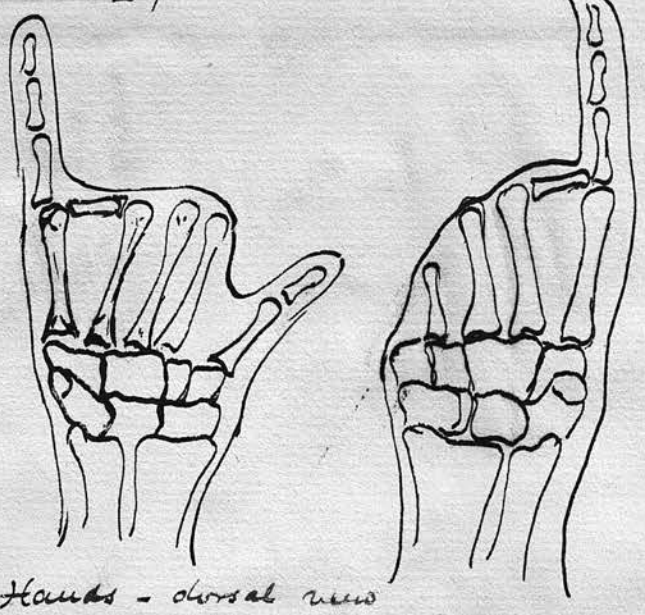
IV. (8) Henry W - hands and feet defective. Hands said to have ~~9 1/2~~ digits only on each (which not specified) feet resemble those of his sister. (the preceeding case.)

On examination the hands showed the ~~outer two~~ 4th & 5th metacarpals, & possibly the 3rd in a Rudimentary form Ta pezium & Ta pezoid bones probably about. little finger, only, present, having 3 phalanges. Feet as in previous case.

Henry Whitehorn
IV . 8.

Fred Whitehorn
IV . 9.

Left. Right.



Hands - dorsal view

By Fred. Whitehorn.

IV. 9.

IV. (9) Fred W - . Hands and feet deformed.

The deformity of the hands is difficult to determine, (see diagram) but probably the rigid bar of bone felt running along the free border of the hand is due to a displaced phalanx, or to two phalanges fused and displaced. The hands as in the majority of the cases show poor muscular development and the 5 metacarpals are palpable on the ~~right~~ Left hand.

Hands.

	Left.	Right.
Carpus.	Normal.	Apparently normal.
Metacarpals.	Normal but the middle 3 are deflected towards the thumb.	1st absent. 2nd rudimentary. 3rd " but larger. 4th & 5th of normal length.

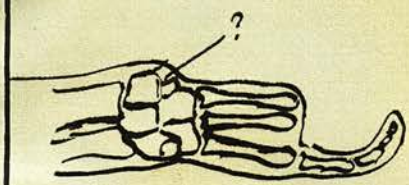
Phalanges.- In both hands - a bone is felt lying between the heads of 4th and 5th metacarpals, probably a phalanx of ring finger.

Left.	Right.
1st digit - rudimentary.	5th digit shows 3 phalanges.
5th " 3 phalanges.	

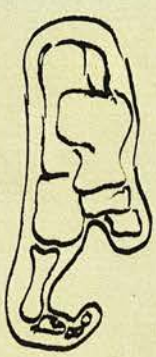
The feet show the same deformity as in Eliza W - his sister.

Wm Whitehorn.

IV. 10.



Right. Hand.



Right. foot.

IV. (10) ~~Thos.~~ Wm Whitehorn.

Hands and feet deformed.

Hands.

Right and left are symmetrical.

Carpals - Trapezium probably absent.

Metacarpals. - The inner three alone present.

Digits. - Little finger only present and has 3 phalanges with freely moveable joints.

Feet.

Also symmetrical.

Tarsus. - The external and middle cuneiform bones are probably absent.

Metatarsals. - 5th alone present.

Digits. - A small and very much curved little toe only is present, and probably has normal number of bones in it.

IV. (12) ~~Reginald W~~ Thos. Wm Whitehorn.

Hands resemble those of his brother IV (10) in every respect.

Feet.

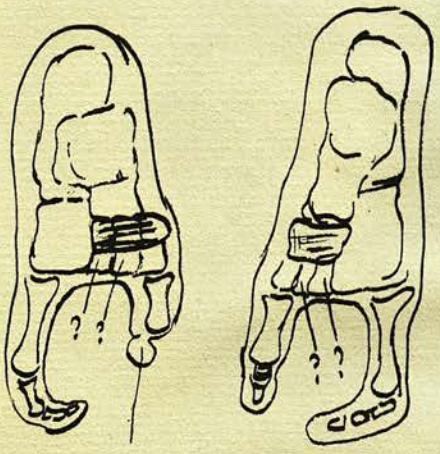
	Right.	Left.
Tarsus.	The presence of the mid and ext. cuneiform bones is doubtful.	Apparently normal.
Metatarsals.	5th alone present. 1st Rudimentary "	~~1st and~~ 5th present.
Digits.	5th alone present and curved.	~~1st one rudimentary phalanx~~. 5th normal in number of bones, but curved.

This child is only 3 years of age and it is difficult to say with any degree of certainty what bones are present and what absent in the case of the carpus, tarsus and the metacarpal and metatarsal bones. This is true also of the following case.

Reginald. Whitehorn. aet. 5.

IV 11.

Feet

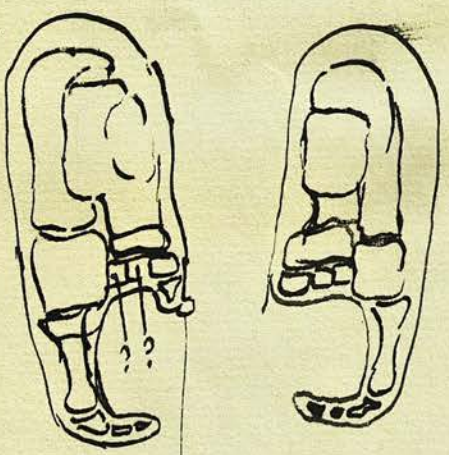


Small fibrous mass.

Thos. W. Whitehorn. aet 3

IV . (12) . (Page. 16)

Feet.



Small mass of fibrous tissue

IV. (1) Reginald Whitehouse

Hands and feet deformed.

The hands are symmetrical and probably have the trapezium, at least, absent. Either the inner 2 or 3 metacarpal bones are present, but are badly developed, except the innermost. One digit, the little finger, is alone present, and has the usual 3 bones, it is very much curved.

The feet are assymetrical.

	Right.	Left.
Tarsus.	- Possibly normal, though the cuneiform bones are not palpable.	
Metatarsals.	1st rudimentary. 5th normal.	1st and 5th present.
Phalanges.	3 bones in 5th digit.	The first toe is long, and apparently composed of two bones. The little toe is as long as a child's middle finger, perfectly straight and without a nail.

The deductions I have arrived at from a study of this family are the following : -

(1) Mendels theory of Dominants and Recessives and Galton's theory of Ancestral Inheritance are equally applicable to the human species.

(2) Though the deformities vary in many cases; in the majority of instances the most highly specilised portions of the hands and feet are deficient. The most highly specilised organs of the body usually are the latest to fully develop - for example, the central nervous system etc. Whether there is any appreciable interval between the appearances of the inner and outer parts of the hand in embryo, I am not in a position to state, but the analogy is interesting.

(3) There is no other developmental defect, save that of the extremities in the affected cases, none the less many of them are paupers, and the inmates of Workhouses. It is obvious that such a defect must seriously interfere with these persons obtaining a livelihood, and though no doubt by a process of natural selection, the deformity will disappear in course of time, still it seems to me, for reasons of national economy, an argument for compulsory sterilisation of the unfit, whether physically or mentally deficient.